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NINDS Fabry's Disease Information Page

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What is Fabry's Disease?
Fabry disease is a fat storage disorder caused by a deficiency of an enzyme involved in the biodegradation of lipids. The gene that is altered in this disorder is on the X-chromosome, so only the mother needs to be a carrier to produce an affected child. Her sons have a 50 percent chance of having the condition, and her daughters have a 50 percent chance of being a carrier. Some of the female carriers exhibit signs of the condition, especially cloudiness of the cornea. In addition to the eye manifestations, males characteristically have burning sensations in their hands and feet that is worse with exercise and hot weather. Most of the males have small, raised, reddish-purple blemishes on their skin. As they grow older, they may have impaired arterial circulation leading to early heart attacks and strokes. The kidneys become progressively involved, and many patients have required kidney transplantation or dialysis. A number of patients have gastrointestinal difficulties characterized by frequent bowel movements shortly after eating. This disorder is due to a deficiency of a lipid breakdown enzyme known as *ceramidetrihexosidase,* also called *alpha-galactosidase A*. Its function is to cleave to a molecule of galactose from a lipid that arises primarily from old red blood cells.

Is there any treatment?
The pain in the hands and feet usually responds to medications such as Tegretol (carbamazepine) and dilantin. Gastrointestinal hyperactivity may be treated with metoclopramide or Lipisorb® (a nutritional supplement). Recent experiments indicate that enzyme replacement is effective therapy for patients with this disorder.

What is the prognosis?
Patients with Fabry disease usually survive into adulthood, but they are at risk for strokes, heart attacks, and kidney damage. It is anticipated that enzyme replacement and eventually gene therapy will eliminate these difficulties.

What research is being done?
NINDS supports research to find ways to treat and prevent lipid storage disorders such as Fabry disease.

Select this link to view a list of studies currently seeking patients.

Organizations

Fabry Support & Information Group
108 NE 2nd Street

P.O. Box 510
Concordia, MO 64020-0510
fabry@fabry.org
http://www.fabry.org
Tel: 660-463-1355
Fax: 660-463-1356

Association for Neuro-Metabolic Disorders
c/o Cheryl Volk
5223 Brookfield Lane
Sylvania, OH 43560
VOLK4OLKS@aol.com
Tel: 419-885-1497

National Tay-Sachs and Allied Diseases Association
2001 Beacon Street
Suite 204
Boston, MA 02135
info@ntsad.org
http://www.ntsad.org
Tel: 617-277-4463 800-90-NTSAD (906-8723)
Fax: 617-277-0134

National Organization for Rare Disorders (NORD)
P.O. Box 1968
(55 Kenosia Avenue)
Danbury, CT 06813-1968
orphan@rarediseases.org
http://www.rarediseases.org
Tel: 203-744-0100 Voice Mail 800-999-NORD (6673)
Fax: 203-798-2291

Provided by:
The National Institute of Neurological Disorders and Stroke
National Institutes of Health
Bethesda, MD 20892

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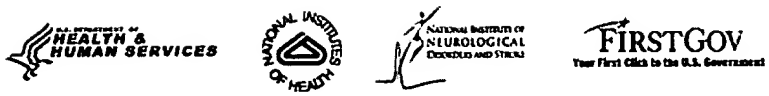
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L2: Entry 4 of 5 File: USPT Aug 14, 2001

DOCUMENT-IDENTIFIER: US 6274597 B1
TITLE: Method of enhancing lysosomal .alpha.-Galactosidase A

Brief Summary Text (15):
Other specific competitive inhibitors for .alpha.-galactosidase, such as for example, calystegine A.sub.3, B.sub.2 and B.sub.3, and N-methyl derivatives of these compounds should also be useful in the methods of the invention. The calystegine compounds can be represented by the formula ##STR2##

Brief Summary Text (16):
wherein for calystegine A.sub.3 : R.sub.1 =H, R.sub.2 =OH, R.sub.3 =H, R.sub.4 =H; for calystegine B.sub.2 : R.sub.1 =H, R.sub.2 =OH, R.sub.3 =H, R.sub.4 =OH; for calystegine B.sub.3 : R.sub.1 =H, R.sub.2 =H, R.sub.3 =OH, R.sub.4 =OH; for N-methyl-calystegine A.sub.3 : R.sub.1 =CH.sub.3, R.sub.2 =OH, R.sub.3 =H, R.sub.4 =H; for N-methyl-calystegine B.sub.2 : R.sub.1 =CH.sub.3, R.sub.2 =OH, R.sub.3 =H, R.sub.4 =OH; and for N-methyl-calystegine B.sub.3 : R.sub.1 =CH.sub.3, R.sub.2 =H, R.sub.3 =OH, R.sub.4 =OH.

Brief Summary Text (22):
or a compound selected from the group consisting of 2,5-dideoxy-2,5-imino-D-mannitol, .alpha.-homonojirimycin, 3,4-diepi-.alpha.-homonojirimycin, 5-O-.alpha.-D-galactopyranosyl-.alpha.-homonojirimycin, 1-deoxygalactonojirimycin, 4-epi-fagomine, and 1-Deoxy-nojirimycin and their N-alkyl derivatives, will alleviate the symptoms of Fabry disease by increasing the activity of mutant .alpha.-Gal A in patients suffering from Fabry disease. Other competitive inhibitors of .alpha.-Gal A, such as calystegine compounds and derivatives thereof should also be useful for treating Fabry disease.

Other Reference Publication (4):
Goldmann et al., "Biological Activities of the Nortropane Alkaloid, Calystegine B2, and Analogs: Structure Function Relationships", J. Natl. Prod., vol. 59, pp. 1137-1142, 1996.*